

This sSMC (X) includes XIST region, allowing the inactivation of this chromosome. Once that it is very small being all the short arm and part of the long arm absent, it is expected that it is preferential inactivated instead of occurring random inactivation. This is in accordance with the absence of ultrasound abnormalities. Nevertheless, the inactivation pattern is not predictable. For the better characterization of this kind of sSMC (X), aCGH should always be performed allowing a more accurate genetic counseling.

## P-13

### X-LINKED ICTHYOSIS – A METABOLIC ETHIOLOGY FOR “DRY SKIN”

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**Introduction:** X-linked ichthyosis is a keratinization genetic disorder characterized by a generalized desquamation of large, adherent, dark brown scales involving trunk and limbs, but sparing palms and soles. It is often associated with other clinical symptoms, such as cryptorchidism (20%), social communication deficits, attention deficit hyperactivity syndrome (40%) or autism (25%). XLI has an incidence of 1 in 6000 births and differs from other types of ichthyosis by transmission mode, clinical manifestations and age of onset. Biochemically, the disorder is due to deficiency in steroid sulfatase (STS), an enzyme localized in the endoplasmic reticulum and responsible for hydrolysis of cholesterol sulfate to cholesterol. Cholesterol sulfate accumulation in patient's epidermis leads to barrier instability and inhibits the desmosomal degradation which is required for normal desquamation, thereby leading to corneocyte retention.

**Aims:** report the etiological identification of XLI, among all genetic disorders, an entity that shows one of the highest ratios of chromosomal deletions (found in up to 90% of patients).

**Methods:** Diagnosis is based on STS enzymatic activity determination as the fraction of total arylsulfatase C activity which is inhibited by dehydroepiandrosterone sulfate. Patients present undetectable levels of STS activity when compared with normal controls.

**Results:** Since 1984, 28 affected males were diagnosed with XLI, some of them within the same family in three different generations. Ichthyosis was present as the first clinical signal.

**Conclusions:** ICX is usually identified as a disease with mild clinical impact and with satisfactory therapeutic response. However, the accurate diagnosis of this disease is crucial to offer patients and affected families proper guidance, regarding attention deficit hyperactivity with predominantly inattentive symptoms. Prenatal diagnosis is available and would be advocated for those cases which have Xp22.3 larger deletions encompassing neighboring genes. These patients may present mental retardation, or features of X-linked chondrodysplasia punctata, in addition to XLI. Severe XLI forms may thus represent contiguous gene deletion syndromes.